



## ALG12 gene

ALG12, alpha-1,6-mannosyltransferase

### Normal Function

The *ALG12* gene provides instructions for making an enzyme that is involved in a process called glycosylation. During this process, complex chains of sugar molecules (oligosaccharides) are attached to proteins and fats (lipids). Glycosylation modifies proteins so they can fully perform their functions. Oligosaccharides are made up of many sugar molecules that are attached to one another in a stepwise process, forming a complex chain. The enzyme produced from the *ALG12* gene transfers a simple sugar called mannose to growing oligosaccharides at a particular step in the formation of the chain. Once the correct number of sugar molecules are linked together, the oligosaccharide is attached to a protein or lipid.

### Health Conditions Related to Genetic Changes

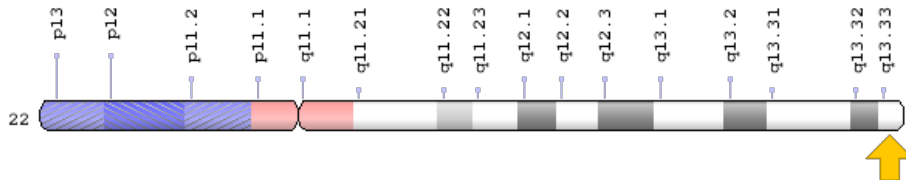
#### ALG12-congenital disorder of glycosylation

At least 13 mutations in the *ALG12* gene have been found to cause *ALG12*-congenital disorder of glycosylation (*ALG12*-CDG). This condition typically leads to delayed growth and development, weak muscle tone (hypotonia), and other signs and symptoms. Mutations in the *ALG12* gene result in the production of an abnormal enzyme with little activity. Without a properly functioning enzyme, mannose cannot be added to the chain efficiently, and the resulting oligosaccharides are often incomplete. Although the short oligosaccharides can be transferred to proteins and lipids, the process is not as efficient as with the full-length oligosaccharide. As a result, glycosylation is reduced. The wide variety of signs and symptoms in *ALG12*-CDG are likely due to impaired glycosylation of proteins and lipids that are needed for normal function in many organs and tissues, including the brain.

## Chromosomal Location

Cytogenetic Location: 22q13.33, which is the long (q) arm of chromosome 22 at position 13.33

Molecular Location: base pairs 49,860,163 to 49,918,469 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- asparagine-linked glycosylation 12 homolog (*S. cerevisiae*, alpha-1,6-mannosyltransferase)
- asparagine-linked glycosylation 12 homolog (yeast, alpha-1,6-mannosyltransferase)
- asparagine-linked glycosylation 12, alpha-1,6-mannosyltransferase homolog
- asparagine-linked glycosylation protein 12 homolog
- CDG1G
- dol-P-Man dependent alpha-1,6-mannosyltransferase
- dol-P-Man:Man(7)GlcNAc(2)-PP-Dol alpha-1,6-mannosyltransferase
- dolichyl-P-Man:Man(7)GlcNAc(2)-PP-dolichol alpha-1,6-mannosyltransferase
- dolichyl-P-Man:Man(7)GlcNAc(2)-PP-dolichyl-alpha-1,6-mannosyltransferase
- dolichyl-P-mannose:Man-7-GlcNAc-2-PP-dolichyl-alpha-6-mannosyltransferase
- ECM39
- hALG12
- mannosyltransferase ALG12 homolog
- membrane protein SB87
- PP14673

## **Additional Information & Resources**

### Educational Resources

- Biochemistry (fifth edition, 2002): Carbohydrates Can Be Attached to Proteins to Form Glycoproteins  
<https://www.ncbi.nlm.nih.gov/books/NBK22521/>

### GeneReviews

- Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview  
<https://www.ncbi.nlm.nih.gov/books/NBK1332>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALG12%5BTIAB%5D%29+OR+%28%28dol-P-Man%5BTIAB%5D%29+OR+%28hALG12%5BTIAB%5D%29+OR+%28dolichyl-P-Man%5BTIAB%5D%29+OR+%28dolichyl-P-mannose%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- ALG12, S. CEREVISIAE, HOMOLOG OF  
<http://omim.org/entry/607144>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ALG12%5Bgene%5D>
- HGNC Gene Family: Dolichyl D-mannosyl phosphate dependent mannosyltransferases  
<http://www.genenames.org/cgi-bin/genefamilies/set/430>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=19358](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19358)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/79087>
- UniProt  
<https://www.uniprot.org/uniprot/Q9BV10>

## Sources for This Summary

- OMIM: ALG12, S. CEREVISIAE, HOMOLOG OF  
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*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1222867/>
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Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/ALG12>

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